



## GYS1 gene

glycogen synthase 1

### Normal Function

The *GYS1* gene provides instructions for making an enzyme called muscle glycogen synthase. Muscle glycogen synthase is produced in most cells but is most abundant in heart (cardiac) muscle and muscles used for movement (skeletal muscles). Muscle glycogen synthase helps link together molecules of the simple sugar glucose to form the complex sugar glycogen, which is a major source of stored energy in the body. Most glucose that is taken in from food is stored as glycogen in muscle cells. During contractions of the cardiac muscle or rapid or sustained movement of skeletal muscle, glycogen stored in muscle cells is broken down to supply the cells with energy.

### Health Conditions Related to Genetic Changes

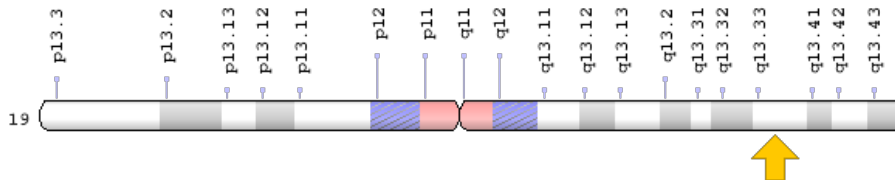
#### glycogen storage disease type 0

At least four mutations in the *GYS1* gene have been found to cause a form of glycogen storage disease type 0 (GSD 0) that affects cardiac and skeletal muscle. Most *GYS1* gene mutations that cause this condition lead to a lack of functional muscle glycogen synthase, resulting in a complete absence of glycogen in muscle cells. Normally, glycogen is formed from the leftover glucose that is not immediately used by cells after glucose is consumed during meals. In people with GSD 0, who cannot form glycogen, the extra sugar is released by the body. As a result, people with muscle GSD 0 do not have any stored energy, which leads to muscle pain, weakness, or episodes of fainting following moderate physical activity. Since there is no glycogen in cardiac muscle, affected individuals are also at an increased risk of cardiac arrest and sudden death, particularly after physical activity.

## Chromosomal Location

Cytogenetic Location: 19q13.33, which is the long (q) arm of chromosome 19 at position 13.33

Molecular Location: base pairs 48,968,125 to 48,993,353 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- glycogen [starch] synthase, muscle
- glycogen synthase 1 (muscle)
- GSY
- GYS
- GYS1\_HUMAN
- muscle glycogen synthase
- muscle glycogen synthase 1

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Glycogen Synthase Catalyzes the Transfer of Glucose from UDP-Glucose to a Growing Chain  
<https://www.ncbi.nlm.nih.gov/books/NBK22413/#A2948>
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/msys/glycogen.html#gsd0>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GYS1%5BTIAB%5D%29+OR+%28glycogen+synthase+1%5BTIAB%5D%29%29+OR+%28%28muscle+glycogen+synthase%5BTIAB%5D%29+OR+%28glycogen+synthase+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- GLYCOGEN SYNTHASE 1  
<http://omim.org/entry/138570>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GYS1.html](http://atlasgeneticsoncology.org/Genes/GC_GYS1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GYS1%5Bgene%5D>
- HGNC Gene Family: Glycosyl transferases group 1 domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/427>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4706](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4706)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2997>
- UniProt  
<http://www.uniprot.org/uniprot/P13807>

### **Sources for This Summary**

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